HHT Outcomes Registry
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Over the course of the last eighteen months, Dr. Marie Faughnan, Cure HHT Scientific Research Director, has been working with collaborators at Geisel School of Medicine at Dartmouth to develop the HHT Outcomes Registry. The registry’s purpose is to collect patient data that will allow scientists to conduct natural history studies as a critical step in furthering research on HHT. In particular, pharmaceutical companies have indicated that access to this patient data resource is a determining factor in their decision to invest financial resources in drug development for HHT. This high priority project is fully funded by Cure HHT through generous donations from members of the foundation.

Excerpts from July 2014 National Patient & Family Conference Presentation by Kristen Anton, Principal Investigator at BioInformatics Service Center at Geisel School of Medicine at Dartmouth

What is a research registry?
A research registry is an organized system that uses observational study methods to collect uniform data to evaluate specified outcomes for a population defined by a particular disease or condition.

Kind of data typically collected includes:
- Patient demographics
- Family history
- Risk factors, exposures
- Disease characteristics
- Disease testing
- Treatment protocols
- Sometimes: biological samples to address genetic characteristics

Why are research registries important?
Research registries provide the ability to:
- Capture standardized, quantifiable information about patients and disease
- Collect population-based data, with enough subjects to scientifically draw conclusions
- Ensure high data quality - so outcomes are reliable
- Collect longitudinal data (sometimes decades)
- Measure or monitor safety and/or effectiveness of treatments
- Springboard new scientific questions

Examples of a successful research registries:
1. Colon Cancer Family Registry (C-CFR)
   - unique resource for investigators to use in conducting studies on the genetics and molecular epidemiology of colon cancer
   - The C-CFR continues to collect data from Registry participants
• Investigators collect information on the patient, the patient’s family, and incidence of cancer. Information from family members both with and without cancer is important
• C-CFR sites span the globe
• Blood and tissue are collected to support ongoing molecular characterization of each family
• 62,353 individuals from 10,662 families
• C-CFR has generated more than 375 peer reviewed publications

2. Infertility Family Research Registry (IFRR)
• helping to improve the health of people and families that have faced a diagnosis of infertility or dealt with infertility treatments
• With 943 participants, the IFRR population has provided enough information to launch three research studies

The HHT Outcomes Registry will include de-identified (privacy protected) clinical, radiographic (X-ray, CT scan, MRI scan), genetic, and treatment information on all HHT patients in North America who are evaluated in HHT Treatment Centers of Excellence and voluntary consent to participate. The data collected by the registry will relate to every aspect of the disease (from nasal bleeding to stroke), with emphasis on determining the outcomes (severity of symptoms, quality of life, heart failure, stroke, severe bleeding, etc. and death), the personal factors that affect these outcomes (genetics, lifestyle, age, sex, etc.) as well as therapies. This information can then be used
to plan and appropriately target clinical research studies of new therapies. By linking all North American HHT Centers, the number of patients is increased twenty times over what any single HHT Center could gather and analyze which makes the Registry a powerful research resource.

We anticipate that a pilot with two HHT Centers will be conducted in Spring 2015. The HHT Outcomes Registry’s short-term goal is to recruit 1000 HHT patients in the first year with 500 data fields per participant while the projected annual recruitment rate is 1,000 per year with an expected maximum of 80,000 patients.

*Funding of this registry is another example of Cure HHT’s goal to transform this disease to advance therapies that are more effective, accelerate discovery, and cure HHT.*